CLAIMS

What is claimed is:

1. An oligonucleotide probe for identifying a mutation in a FIBL-6 gene associated with AMD, said probe comprising a polynucleotide capable of uniquely hybridizing with a polynucleotide comprising a sequence having the nucleotide sequence of positions 16,255 to 16,270 of SEQ ID NO:1, wherein the nucleotide at position 16,263 of SEQ ID NO:1 is mutated.

2. The oligonucleotide probe of claim 1, wherein said probe comprises a polynucleotide selected from the group consisting of SEQ ID NO:8 and 95-177.

3. An oligonucleotide probe for identifying a wild type FIBL-6 gene wherein a mutation in said gene is associated with AMD, said probe comprising a polynucleotide capable of uniquely hybridizing with a polynucleotide comprising a sequence having the nucleotide sequence of positions 16,255 to 16,270 of SEQ ID NO:1.

4. The oligonucleotide probe of claim 4, wherein said oligonucleotide probe comprises a polynucleotide selected from the group consisting of SEQ ID NO:7 and 12-94.

5. The oligonucleotide probe of claim 5, wherein said probe consists of a polynucleotide selected from the group consisting of SEQ ID NO:7 and 12-94.

6. A kit for identifying a polynucleotide mutation associated with AMD, said kit comprising the probe of any one of claims 1-3 and 7, and reagents for hybridizing said probe to said polynucleotide.

7. A method of determining whether a subject is at risk for development of macular degeneration, the method comprising the steps of:

(a) obtaining a nucleic acid sample from the subject; and

(b) conducting an assay on the nucleic acid sample to determine the presence or absence of a FIBL-6 gene mutation associated with macular degeneration, wherein the presence of a FIBL-6 gene point mutation associated with macular degeneration indicates that the subject is at risk for development of macular degeneration.

8. The method of claim 7, wherein the assay is selected from the group consisting of probe hybridization, direct sequencing, restriction enzyme fragmentation and fragment electrophoretic mobility.

9. The method of claim 7 wherein the nucleic acid sample is an RNA sample and the assay is a direct sequencing assay.

10. The method of claim 9, wherein the assay comprises the steps of:

(a) reverse transcribing the RNA sample to produce a corresponding cDNA;

(b) performing at least one polymerase chain reaction with suitable oligonucleotide primers to amplify the FIBL-6 cDNA;

(c) obtaining the nucleotide sequence of the amplified FIBL-6 cDNA; and

(d) determining the presence or absence of a FIBL-6 gene point mutation.

11. The method of claim 10, wherein step (d) comprises determining the presence or absence of a mutation of the polynucleotide of SEQ ID NO:1, the mutation being a substitution of at least one base of the codon at position 16,262, 16,263 and 16,264 wherein said mutated codon does not encode Gln.

12. The method of claim 11, wherein said mutation produces a codon for arginine.

13. The method of claim 7 wherein the nucleic acid sample is a DNA sample.

14. The method of claim 9 wherein the DNA sample is a genomic DNA sample and the assay comprises the steps of:

(a) amplifying a target portion of the nucleotide sequence of the genomic DNA;

(b) obtaining the nucleotide sequence of said amplified portion;

(c) determining the presence or absence of a FIBL-6 gene mutation associated with macular degeneration in said target portion nucleotide sequence.

15. The method of claim 14, wherein step (c) comprises determining the presence or absence of a mutation of the polynucleotide of SEQ ID NO:1, the mutation being a substitution of at least one base of the codon at position 16,262, 16,263 and 16,264 wherein said mutated codon does not encode Gln.

16. A method for determining whether a subject displaying symptoms is suffering from familial AMD, the method comprising the steps of:

(a) obtaining a nucleic acid sample from the subject; and

(b) conducting an assay on the nucleic acid sample to determine the presence or absence of a FIBL-6 point mutation associated with AMD, wherein the presence of a FIBL-6

point mutation associated with AMD indicates that the subject is suffering from AMD.

17. A method for determining whether a subject is free of AMD associated with a mutation of the FIBL-6 gene, the method comprising the steps of:

(a) obtaining a nucleic acid sample from the subject; and

(b) conducting an assay on the nucleic acid sample to determine the presence or absence of a FIBL-6 gene point mutation associated with AMD, wherein the absence of a FIBL-6 point mutation associated with AMD indicates that the subject is free of AMD associated with a missense mutation of the FIBL-6 gene.

18. The method of claim 17, wherein the nucleic acid sample is an RNA sample and the assay is a direct sequencing assay.

19. The method of claim 17, wherein the nucleic acid sample is a DNA sample.

20. The method of claim 17, wherein the nucleic acid sample is a genomic DNA sample and the assay comprises the steps of:

(a) amplifying a target portion of the nucleotide sequence of the genomic DNA;

(b) obtaining the nucleotide sequence of said amplified target portion; and

(c) determining the presence or absence of a FIBL-6 gene point mutation associated with AMD in said target portion nucleotide sequence.